Practice

A Patient's Journey

External aortic support for people with Marfan's syndrome

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In 2007 Camilla Allen became the ninth person with Marfan's syndrome to have an external aortic root support wrapped around her dilated aorta. She has since become the first woman with an exostent to become pregnant and give birth.

Stopped in my tracks

In October 2006 I agreed to take part in research related to Marfan's syndrome being undertaken at St George's Hospital in London. The research involved detailed measurements of my heart being taken using echocardiograms. I did not give my participation a second thought until a couple of months later when I received a letter from St George's telling me that my aortic root diameter had expanded to 4.3 cm and that with such a dilated aorta I should not conceive a child due to the risk of fatal aortic dissection. This news was a devastating blow to my husband and me as we had been trying to conceive our second child for the previous nine months.

An appointment with my local cardiology consultant was hastily arranged for January 2007. The measurement was confirmed, and, although it was not particularly worrying in itself, as most patients undertaking root and valve replacement surgery have aortic root diameters nearer the 5.0 cm mark, it showed that mine had increased since my first echocardiogram at the age of 14 and had worsened during my first pregnancy. The consultant considered surgery in the form of the Bentall procedure, a composite aortic root replacement, which is a necessary precaution against a 1 in 10 chance of aortic dissection during pregnancy or labour. Suddenly a 10% chance of my dying brought the situation into sharp focus.

Since first being diagnosed with Marfan's syndrome as a teenager, I had always considered my diagnosis to be an inconvenience rather than life threatening. The condition is only mild in my family, so it hasn't dominated my life. I had annual echocardiograms, and, although my aortic root diameter had steadily increased from 3.3 cm at age 14 to 4.3 cm at the age of 35, I felt fit, healthy and in no immediate danger. With only minor heart monitoring I had given birth naturally to one child already. It was therefore very difficult to stomach the need for major aortic valve heart surgery with all that it entails.

I am no stranger to surgical procedures, so such a major operation did not fill me with dread—especially as it was a well established procedure with excellent success rates. However, I did not relish the idea of undergoing major heart surgery that included the need for cardiopulmonary bypass and for the removal of my healthy aortic tissue. I was also particularly concerned and frustrated that I would need to have blood thinning anticoagulants for life and that I would need to inject these anticoagulants if I became pregnant. Despite my reservations I was beginning to

think it was a foregone conclusion and that there was no alternative. The Henry Ford quote "You can have [a Model T in] any colour as long as it's black" sprang to mind.

Light at the end of the tunnel

My parents had always encouraged me to choose my own path in life and accept responsibility for my decisions. I have therefore become an independent, decisive, and optimistic person who makes decisions based on all the available information. I did not regard this situation as any different and therefore refused to believe that there was only one way forward.

I discussed the matter with my husband, and we decided to seek a second opinion. Thankfully another consultant who specialised in Marfan's syndrome pointed us towards Professor John Pepper at the Royal Brompton Hospital in London. Within a week we were sitting in front of him, and he agreed that we needed to act quickly. He explained the option of the exostent procedure, how it worked, and why he thought I was an ideal candidate for it. I said immediately that I wanted to be considered. Putting a bespoke stent around my aorta, which was produced using magnetic resonance imaging data and computer assisted design, seemed logical and sensible. Firstly it meant leaving my perfectly healthy (albeit a bit stretched) aortic tissue and valve intact, and it had the added benefits of having a shorter recovery period and no need for me to take anticoagulation medication for the rest of my life.

My husband and I discussed the pros and cons of the Bentall and exostent procedures. Even though it was new, the exostent option still seemed a win-win situation. Because of its less invasive nature it could be reversed if for some reason it was not successful, and the fallback position would be the Bentall procedure. Time was of the essence, and I therefore asked to be operated on as soon as possible with no concern that only eight exostent operations had taken place to date. Someone had to be the ninth: why not me?

Within a few weeks, in March 2007, I was on the operating table having an exostent successfully implanted. The treatment at the Brompton was incredibly stress free and of a very high standard. After only five days in hospital I was home, and within a couple of months, I felt like my old self again, but I now had a healthy and secure aortic root measurement of 3.6 cm.

Back on track

Within a few months of the operation I was allowed to try for another baby, and in November 2007 I became pregnant again. Being the first person to become pregnant after an exostent operation, I was closely monitored in my home town of Bristol. This meant regular echocardiograms and a magnetic resonance imaging scan at 22 weeks' gestation to check that my exostent was holding well. Five days before the due date my daughter decided it was time to make her entrance, and I gave birth in August 2008. I was treated as high risk in the maternity ward but was fortunate to have a labour that was quick, natural, and very unremarkable. My blood pressure was closely monitored before and after the birth but never rose particularly high. Relieved consultants arranged an echocardiogram and a magnetic resonance imaging scan after the birth, and they proved that the exostent had done what it was designed to do. My aortic root measurement remained stable at 3.6 cm.

On reflection

As of October 2009 the exostent procedure has been carried out on 20 people with Marfan's syndrome and has been accepted as a clinically approved procedure at the Royal Brompton Hospital.

For myself, this innovative procedure was the ideal solution to my predicament, but it has always frustrated me that I came so close never to hearing about it. It was only my "refuse to accept it" mentality that led me to seek a second opinion and so discover the alternative course of action. My concern is that some professionals who work with people with Marfan's syndrome choose, for whatever reason, not to present the exostent as an alternative to the Bentall operation. Of course it does not yet have a long history of success, and not all patients would be suitable. Others may make different choices. However, for those who have healthy aortic tissue and valves, the exostent can offer a less drastic alternative to the Bentall procedure.

I must confess to not having known much about the procedure before my operation, but since then I have learnt a lot more about its conception and application. In addition, I have met and enjoyed the ongoing support and friendship of the key players involved in the development of the exostent, Tal Golesworthy and Tom Treasure. They are publicising the exostent technique, especially in the Marfan's syndrome community, and are making sure that research is ongoing. As part of this I am involved in a mentoring scheme for possible recipients of an exostent, which goes some way to helping publicise the procedure. I hope that this will ensure that all people with Marfan's syndrome receive the necessary information and support to make informed decisions when embarking upon a journey similar to mine.

Box 1 A doctor's perspective

In the third trimester, pregnancy enhanced levels of progesterone help to soften the ligaments of the pelvis to ease the path of the baby during delivery. Unfortunately in the presence of a weakened aorta associated with Marfan's syndrome there is a risk that the aorta may enlarge and rupture. The current standard approach is to perform the Bentall operation. This is a safe and reliable procedure, but, as Camilla says, it involves cardiopulmonary bypass and replacement of the aortic valve. The valve substitute can be mechanical, which is durable but necessitates lifelong anticoagulation and is an extra hazard for pregnancy, or it can be a tissue valve, which does not require anticoagulation but is of limited durability.

We wanted to prevent further expansion of the aortic root in the simplest way possible. Wrapping the aorta with artificial material is an old idea, but the suggestion by Golesworthy of using modern computer aided design to produce a bespoke sleeve seemed very attractive. We have since developed an individually tailored external "jacket" for the aortic valve, aortic root, and ascending aorta for patients with Marfan's syndrome.

By using digital information from magnetic resonance imaging, computer aided design and rapid prototyping we are able to create a replica of the patient's aorta. By heat shrinking an appropriate piece of medically approved plastic cloth, Dacron, we can produce a jacket that exactly matches the contours of the patient's aorta, including the sinuses of Valsalva and the origins of the coronary arteries.

This jacket is sterilised and taken to the operating room where, under a general anaesthetic and a midline sternotomy, it is placed around the patient's aortic root and ascending aorta. There is no need for cardiopulmonary bypass or a period of global myocardial ischaemia, and 19 of these operations have now been successfully performed.

Camilla's sleeve was fitted by a relatively short and simple operation. The effect of the exostent on the

diameter of her aorta during the second pregnancy compared to the first is remarkable (fig ↵).

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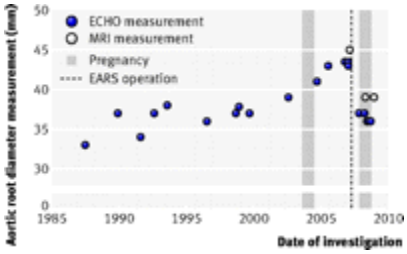

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Fig Aortic root diameter measurement (mm). Image courtesy of Martin Utley and Tom Treasure, Clinical Operational Research Unit, University College London

Box 2 Further information
The Marfan Trust (www.marfantrust.org)—Funds research into the cause and possible prevention of Marfan's syndrome, educates patients and doctors about the condition, and assists with patient support

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This is one of a series of occasional articles by patients about their experiences that offer lessons to doctors. The *BMJ* welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance

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